CHILDREN'S TUMOR FOUNDATION
ENDING NF THROUGH RESEARCH

NF Diagnosis: **Genetic Testing in NF**

This resource reviews the option of genetic testing for all types of neurofibromatosis and schwannomatosis when a diagnosis is suspected or when confirmation of the genetic cause of a condition is recommended. This information does not provide medical advice or replace information provided by your healthcare provider.

Genetic testing is available for all forms of NF, which refers to neurofibromatosis type 1 (NF1) and all types of schwannomatosis (SWN), including *NF2*-related schwannomatosis (*NF2*-SWN).

WHAT IS GENETIC TESTING?

Genetic testing, also called DNA or molecular testing, involves evaluating one or more genes. Variants (previously called mutations) are changes in a gene that may disrupt the gene's function. If a gene's function is significantly changed, it can lead to a medical condition such as NF1 or a type of SWN.

WHAT IS INVOLVED IN GENETIC TESTING?

Most genetic testing is performed on blood or saliva. In some situations, testing of skin or another area of the body is required. Results of genetic tests are typically available in 1-2 months.

WHO SHOULD CONSIDER GENETIC TESTING?

Genetic testing can be helpful in a variety of situations:

- To confirm a diagnosis
- To provide information about the severity or likelihood of specific features (in some cases)
- To identify other family members with the condition
- To allow for the consideration of reproductive options such as preimplantation or prenatal testing (see CTF resource: Family Planning and Reproductive Options in NF)

WHAT ARE THE POSSIBLE RESULTS OF GENETIC TESTING?

For each gene evaluated, there are three possible results.

1. **The test detects a genetic change known to cause the condition (pathogenic variant).**
   A genetic variant is identified and confirms the diagnosis in an individual.
2. **The test does not detect a genetic variant.**
   A genetic variant causing the condition is not identified in the gene tested. This is sometimes called a negative result. A negative NF1 or SWN genetic test result should be interpreted with caution by a medical provider familiar with the nuances of genetic testing in these conditions. A negative result may mean that an individual does not have the condition. However, there are other reasons for a negative test in an individual who has the condition, including the laboratory method used for the testing or the type of body tissue tested.
3. **The test detects a variant of uncertain significance (VUS).**
   A genetic variant was detected, but the variant is not definitively associated with causing the condition. This is an inconclusive result called a variant of unknown significance. Testing of additional family members may be done to assist in the interpretation of the results.

   New technologies continue to be developed as we learn new information about all types of NF, so improved testing and interpretation of results is anticipated in the future.

WHAT ARE OTHER CONSIDERATIONS OF GENETIC TESTING?

There are some limitations to genetic testing.

- **If a pathogenic variant is identified**, genetic testing often does not predict the severity of the condition or determine with certainty how an individual will be impacted.
- **If no variant is identified**, a diagnosis is not excluded.
  - Additional testing might be recommended by different lab techniques or in different body tissue.
  - If someone is highly suspected of having a condition, a negative genetic test might not change the medical screening recommendations.
  - If a diagnosis is known based on clinical features, a negative genetic test does not change the diagnosis.
- **If a VUS is identified**, additional testing of family members is often requested but may not help in the final interpretation of the result.

Genetic testing is expensive, and a medical insurance plan may or may not cover the costs. You should check with your insurance company or healthcare provider to determine what out-of-pocket expenses may be associated with the testing.

WHAT IF YOU HAVE QUESTIONS ABOUT GENETIC TESTING?

Genetic testing is voluntary, and a family should be aware of the costs, benefits, and limitations before deciding to proceed. Talk with your genetic counselor or healthcare provider for more information about genetic testing.

To locate a genetic counselor or NF specialist near you, visit **ctf.org/doctor**.

For more information about the genetics of NF, visit **ctf.org/genetics**.

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Written by Heather Radtke, MS, CGC, Medical College of Wisconsin
Reviewed by Tena Rosser, MD, Children's Hospital Los Angeles

212-344-6633 | 1-800-323-7938 | info@ctf.org | ctf.org |